



BBS1 gene

Bardet-Biedl syndrome 1

Normal Function

The *BBS1* gene provides instructions for making a protein found in cells throughout the body. The BBS1 protein is part of a group (complex) of proteins that plays a critical role in the formation of cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of many types of cells. They are involved in cell movement and many different chemical signaling pathways. Cilia are also necessary for the perception of sensory input (such as sight, hearing, and smell).

Health Conditions Related to Genetic Changes

Bardet-Biedl syndrome

More than 30 mutations in the *BBS1* gene have been identified in people with Bardet-Biedl syndrome. Mutations in this gene are the most common cause of Bardet-Biedl syndrome, accounting for about one-quarter of all cases.

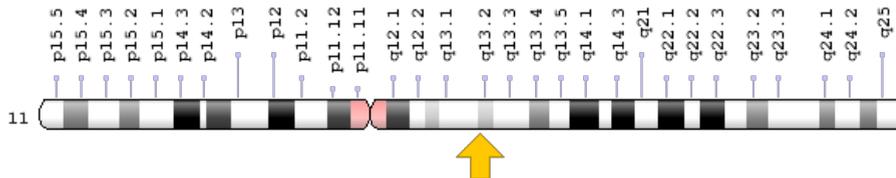
Most *BBS1* gene mutations change single protein building blocks (amino acids) in the BBS1 protein or lead to the production of an abnormally short version of the protein. The most common mutation replaces the amino acid methionine with the amino acid arginine at protein position 390 (written as Met390Arg or M390R).

Mutations in the *BBS1* gene likely affect the normal formation and function of cilia. Defects in these cell structures probably disrupt important chemical signaling pathways during development and lead to abnormalities of sensory perception. Researchers believe that defective cilia are responsible for most of the features of Bardet-Biedl syndrome, including vision loss, obesity, the presence of extra fingers and/or toes (polydactyly), kidney abnormalities, and intellectual disability.

Chromosomal Location

Cytogenetic Location: 11q13.2, which is the long (q) arm of chromosome 11 at position 13.2

Molecular Location: base pairs 66,510,648 to 66,533,613 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BBS1_HUMAN
- BBS2L2
- FLJ23590

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (first edition, 2000): Cilia and Flagella: Structure and Movement
<https://www.ncbi.nlm.nih.gov/books/NBK21698/>

GeneReviews

- Bardet-Biedl Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1363>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28BBS1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- BBS1 GENE
<http://omim.org/entry/209901>

Research Resources

- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=BBS1%5Bgene%5D>
- HGNC Gene Family: Bardet-Biedl syndrome associated
<http://www.genenames.org/cgi-bin/genefamilies/set/980>
- HGNC Gene Family: BBSome
<http://www.genenames.org/cgi-bin/genefamilies/set/1122>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=966
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/582>
- UniProt
<http://www.uniprot.org/uniprot/Q8NFJ9>

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